

South Dakota Newborn Screening Advisory Committee

April 28, 2022 10:00am – 12:00pm CT Tieszen Building – Basement Level Conference Room and Zoom 306 E Capitol Ave. Pierre, SD 57501

Members Present	Members Absent	Others Present
South Dakota Department of Health	South Dakota Department of Health	State Hygienic Laboratory at
Beth Dokken, Division Director Family &	Dr Mary Carpenter, Medical Director	University of Iowa
Community Health		Stanton Berberich, Program
Linda Ahrendt, Administrator, Office of	Monument Health	Manager
Child & Family Services	Kelli Sorenson, Lab Manager	Travis Henry, Assistant Research
Jennifer Folliard, Assistant Administrator,	Nan Fitzgerald, NICU Manager	Scientist
Office of Child & Family Services		Tate Kappell, Clinical Lab
Lauren Pierce, Newborn Screening	Parent & Family Representative	Supervisor
Coordinator	Jackie Vander Woude, Avera NICU	Carol Johnson, Newborn Screening
Bernadette Boes, Newborn Screening Coordinator	Family Support	Follow up Program Coordinator
Sarah Aker, South Dakota Medicaid		SD EHDI Coordinator
		Shelby Hintze Jepperson, USD
Sanford Health		South Dakota Hands and Voices
Sanford Children's Hospital NBS Medical		Jill Scott
Consultant Genetics/Metabolic		
Dr. Laura Davis-Keppen		Other Public
Dr. Isum Ward		Miranda McAuliffe, ALD Alliance
Indian Health Services		
Dr Carmen Ruiz, Pine Ridge, Pediatric		
Clinic		
Avera Health		
Alyssa Christensen, OB Nurse Manager		

Agenda Item	Discussion
Welcome and Introductions	 Beth Dokken began the meeting at 10:00 am. Meeting took place in person & zoom Introductions from SD DOH, committee members, and presenters.
Purpose of the Committee and Role of the Committee Members	 Jennifer Folliard provided overview of the purpose of the Newborn Screening Advisory Committee (NBSAC). The committee is intended to be advisory in nature meant to advise the South Dakota Newborn Screening (SD NBS) Program. Future meeting topic: Development of Purpose Statement for South Dakota to help focus our efforts and communicate with the public. Bernadette Boes, RN newest member of the SD newborn screening program was introduced. Her background was provided and is currently working with Lauren Pierce current coordinator in learning the role as Lauren is transitioning. Lauren will be leaving the role in August 2022 to return to further her education.
South Dakota Newborn Screening Program	 Bernadette Boes provided a presentation on the overview SD newborn screening bloodspot program, and the purpose and responsibilities of the state of South Dakota. The daily duties of the coordinator and follow up nurse were discussed. Numerical data reviewed number screened, follow up cases and positive screens. Lauren Pierce reviewed Sharina's follow up program with a case study.
Spinal Muscular Atrophy (SMA) Screening Update	 SMA Screening began 9/1/21 no positives as of 4/19/21- 7561 babies have been screened Average prevalence US 1:11,000 lowa 1:6,988, ND 1:21,500 Travis Henry explained that the lab assay is very specific to an exon 7 deletion within SMN1. (95% of affected individuals) He explained that deletion in the Hutterite population encompasses the same region as SMN1 exon 7.
Laboratory & Initial Follow-Up Services RFP Process	 Lauren Pierce reviewed the RFP process October 2021: Released RFP Pompe Disease Screening request included in RFP December 2021: Proposals due 2 proposals received January – February 2022: Evaluation process Diverse evaluation team conducted in-depth evaluation March 2022: Selected successful proposal: State Hygienic Laboratory at the University of Iowa Pompe Disease Screening – Next steps: Summer 2022 - Attend legislative Interim Rules Review Committee to prepare for Pompe disease screening

	Planning and assay update from Iowa State Lab
Pompe Disease Screening Planning Update	 Tate Kappell explained the assay will measure the GAA enzyme 1st tier screen to measure 3 analytes, looks for an enzyme deficiency 6 enzyme test will be run if enzyme deficiency present to look for environmental factors ex. Heat If GAA is still decreased but other 6 are not, rescreen will be requested Second tier screening is still in progress
Enhancing Disease Detection in Newborns Grant Overview Iowa State Hygienic Lab	 Iowa's plan is to add Pompe, MPS 1, and X-ALD by July 1, 2023. Possibly MPS II. The Grant would be used to fund these disorders developmental testing, if awarded. Adding any new condition to a state panel takes resources (money, personnel, space, etc.) Need to purchase Automated liquid handler
MPS I, MPSII and X-ALD Overview	 Dr. Laura Davis Keppen & Dr. Isum Ward gave an overview and history of Newborn bloodspot screening, goals, criteria, RUSP, and conditions on the RUSP that are not yet being screened in SD. Not Screened in SD – X-ALD, MPS I, MPS II MPS I lack treatment can lead to death; successful treatment reviewed Masonic Children's Hospital in MN has very effective treatment in our region. Dr Ward has a close working relationship with them X-ALD disease causes irreversible dementia ultimately leading to death, successful treatment has a critical time window, with treatment curative
Committee Discussion and Recommendations	 Beth Dokken highlighted all the progress and good work that has happened in the past year since our first meeting. She commended everyone on their commitment and input. She offered her thanks and gratitude to the group and she looks forward to the continued progress in our state. She asks that if you have additional input you would like to hear during these meetings that Bernadette Boes is the new point of contact. Reach out to her with suggestions for future meetings at bernadette.boes@state.sd.us. Dr. Ward: In the past, have been uncertain about when to involve Child Protection Services (CPS). Seemed to be confusion amongst hospital/healthcare staff. Potential for communication between services between our program and families? If babies screen positive, sometimes there is resistance from families to do follow-up testing. How do we combat this? Sarah Aker with SD Medicaid said she can take that back and have discussion with CPS Linda asked about barriers to follow-up care. Dr. Ward has had families who have not shown up to appointments.

- Worried that families will not understand importance of follow-up.
- Dr. Davis-Keppen said they have seen this with PKU.
 Families not following treatment until condition gets dangerous.
- Do we know why parents are hesitant to do follow-up treatment/care?
 - Cost
 - Follow-up appointment no-shows despite multiple attempts
 - At what point and in what situation should CPS become involved?
- Sarah Aker: Would be helpful to have added information about efficacy and outcomes of treatments for disorders Dr. Ward and Dr. Davis -Keppen covered. We will take that back and consider that as we develop agenda for future meetings.
- Beth Dokken: Does SD Medicaid cover treatment for any of these new conditions (MPS I, MPS II, X-ALD)? Another important consideration.
 - Dr. Davis- Keppen . These are similar to other things that are already on the newborn screening panel.
 - Children have been covered by Medicaid for these treatments in the past.
 - Cost associated with being involved in a clinical trial are generally covered by Medicaid but if drug is not approved by FDA, Medicaid will not cover it.
 - Not sure if gene therapy is covered by Medicaid. Not essential for a cure.
 - Beth recap: Medicaid does typically cover the therapy.
 Medicaid doesn't typically cover enzyme therapy unless
 FDA approved. Dr. Davis-Keppen indicated that most therapies have been FDA approved. There may be one drug for X-ALD that has not been approved. Something to continue to discuss.
 - Miranda (chat): ALD gene therapy review coming from FDA soon
- Dr. Davis-Keppen: Newborn hearing screening. Many children are identified with hearing loss late and it impacts language. Follow-up is not done with hearing in SD. Ensure that parents get appropriate education and that children get additional follow-up care. At least 1/3 are not getting diagnosed until later. She was advised DOH works with USD through the HRSA grant to improve testing.

Public Comment Opportunity	Miranda Mc Auliffe , ALD Alliance: Mother of a son who diagnosed	
	with X-ALD in NY 6 years ago. She is a partner with Elisa Sieger at	
	ALD Alliance who advocate for ALD to be added to state newborn	
	screening programs. Elisa's son was diagnosed with ALD after	
	lengthy diagnostic tests for months and has since passed away.	
	Elisa turned her grief into advocacy and passion when she learned	
	that there was a NBS for ALD. She started the ALD Alliance. Miranda	
	joined the group when her son received this difficult diagnosis.	
	Miranda learned of the diagnosis through newborn screening	
	thanks to Elisa efforts in her state. Elisa's son is happy and healthy	
	6-year-old boy. Many other of her own family members have been	
	diagnosed and/or are carriers. They have been able to make	
	educated decisions about growing families. ALD Alliance (created by	
	Elisa) created a parent guide for ALD. Roadmap for families new to	
	the diagnosis. Started a free care-package for ALD families. Running	
	an ALD NBS consortium. She provided the following web sites	
	 https://investor.bluebirdbio.com/news-releases/news-release- 	
	details/bluebird-provides-update-fda-review-timelines-	
	betibeglogene	
	https://www.aldalliance.org/family-support.html	
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	Beth Dokken: We will keep everyone updated via email with Pompe	
'	testing and moving through the administrative rule process.	
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Discuss Next Meeting	Next SD NBSAC meeting – Our plan for these meetings going forward: We	
	will continue hybrid model. Will meet annually in the spring but will put	
	together the meeting earlier if needed	